



ADAMTS10 gene

ADAM metallopeptidase with thrombospondin type 1 motif 10

Normal Function

The *ADAMTS10* gene provides instructions for making an enzyme that is found in many of the body's cells and tissues. This enzyme is part of a family of metalloproteases, which are zinc-containing enzymes that cut apart other proteins. Although the function of the *ADAMTS10* enzyme is unknown, it is critical for growth before and after birth. Researchers believe that it may be involved in the development of structures including the skin, eyes, heart, and skeleton.

Health Conditions Related to Genetic Changes

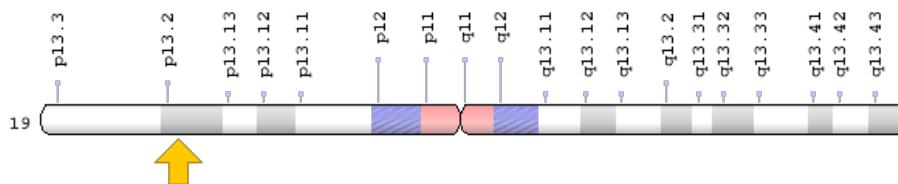
Weill-Marchesani syndrome

At least five mutations in the *ADAMTS10* gene have been identified in people with Weill-Marchesani syndrome. Each of these mutations prevents the cell from producing any functional *ADAMTS10* enzyme. Researchers speculate that a loss of this enzyme disrupts skeletal development, leading to short stature and unusually short fingers and toes (brachydactyly). A shortage of the *ADAMTS10* enzyme also interferes with the development and function of the lens of the eye, causing eye abnormalities and impaired vision. Additionally, a lack of this enzyme may disrupt the normal development of the heart, resulting in the heart defects occasionally seen in people with Weill-Marchesani syndrome.

Chromosomal Location

Cytogenetic Location: 19p13.2, which is the short (p) arm of chromosome 19 at position 13.2

Molecular Location: base pairs 8,580,240 to 8,610,735 on chromosome 19 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- a disintegrin and metalloproteinase with thrombospondin motifs 10
- a disintegrin-like and metalloprotease (reprolysin type) with thrombospondin type 1 motif, 10
- a disintegrin-like and metalloprotease domain with thrombospondin type I repeats 10
- ADAM metallopeptidase with thrombospondin type 1 motif, 10
- ADAM-TS10
- ADAMTS-10
- ATS10_HUMAN

Additional Information & Resources

GeneReviews

- Weill-Marchesani Syndrome
<https://www.ncbi.nlm.nih.gov/books/NBK1114>

Genetic Testing Registry

- GTR: Genetic tests for ADAMTS10
<https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=81794%5Bgeneid%5D>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28ADAMTS10%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- A DISINTEGRIN-LIKE AND METALLOPROTEINASE WITH THROMBOSPONDIN TYPE 1 MOTIF, 10
<http://omim.org/entry/608990>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_ADAMTS10.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=ADAMTS10%5Bgene%5D>
- HGNC Gene Family: ADAM metallopeptidases with thrombospondin type 1 motif
<http://www.genenames.org/cgi-bin/genefamilies/set/50>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=13201
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/81794>
- UniProt
<http://www.uniprot.org/uniprot/Q9H324>

Sources for This Summary

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